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Effects of heterozygosity for Robertsonian translocations on meiotic segregation and fertilization competence of male germ cells. F. Marchetti, X. Lowe and A.J. Wyrobek. Biology and Biotechnology Research Program, Lawrence Livermore National Laboratory, Livermore, CA.

Robertsonian (Rb) translocations are common in mammals. In man about 5% of individuals with Down syndrome have a Rb translocation. Also, individuals Rb heterozygous have elevated frequencies of unbalanced gametes, and therefore are at high risk for spontaneous abortions and chromosomally imbalanced offspring. To study both meiotic segregation and fertilization competence of sperm produced by Rb translocation carriers, we performed FISH painting analysis of mouse metaphase II (MII) spermatocytes and first-cleavage (1-CI) zygotes in male mice doubly heterozygous for the Rb(6.16) and Rb(16.17) translocations. These mice are used to generate trisomy 16, which is a mouse model for Down syndrome. Fifteen Rb mice were mated with normal B6C3F1 females. 1-CI zygotes were collected from mated females ~18 h after fertilization and prepared for metaphase analysis. Testes were then isolated from four randomly chosen mice and testicular preparations for MII spermatocyte analyses were prepared. Hybridization was performed with a cocktail mixture containing three biotin-labeled probes specific for chromosomes 8, 16 and 17 plus a digoxigenin-labeled probe specific for chromosome Y. The frequencies of aneuploid male-derived complements were very similar before and after fertilization. Balanced male complements were 60.6% (57/94) in MII spermatocytes and 57.8% (63/109) in 1-CI zygotes. Disomy or nullisomy for 16 represented 20.2% and 18.1% in MII spermatocytes and 23.9% and 18.3% in 1-CI zygotes, respectively. Disomy for 17 was 1.1% in MII spermatocytes and 0.9% in 1-CI zygotes. No aneuploidy involving chromosome 8 was found, as expected, while a possible segmental aneuploidy involving the Y was observed in a 1-CI zygote. These results show that Rb translocation carriers are at high risk of producing gametes unbalanced for those chromosomes involved in the translocation and suggest that aneuploid sperm are not selected against during fertilization and the zygotic cell cycle.

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Cytogenetics**6. Application for Student Award Consideration**

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